CONGENITAL ADRENAL HYPERPLASIA (CAH)

What is CAH?

CAH is a group of autosomal recessive genetic conditions. The Newborn Screening Program screens for the most common form of CAH, which is 21-hydroxylase deficiency. This disorder is characterized by an inability to adequately produce the hormones cortisol and aldosterone. The lack of cortisol results in excess production of ACTH and an overproduction of androgens by the adrenal gland. Together, the deficiency of cortisol and aldosterone causes adrenal crisis and can be life threatening.

Screening consists of an Elisa method of testing the 17-hydroxprogesterone (17-OHP) level. 17-OHP is the immediate precursor to the missing enzyme and will be elevated in CAH. Elevated 17-OHP levels can occur in sick or low birth weight infants and therefore normal ranges are stratified based on birth weight. Abnormalities on the first assay trigger a second tier test using the Tandem Mass (MSMS) testing method.

Clinical Signs and Symptoms

Female infants present at birth with ambiguous genitalia — enlarged clitoris, posterior labial fusion, and rogation of the labia. The infant may resemble a male with hypospadius, undescended testes and be discharged from the hospital with an erroneous male gender assignment.

Male infants have normal appearing external genitalia at birth and usually present in the first few weeks of life with dehydration, vomiting, lethargy, poor feeding, and shock. This may be life threatening.

Adrenal Crisis	
Signs:	**Symptoms:**
Lethargy	Hyperkalemia
Poor Feeding and/or vomiting	Hyponatremia
Excessive weight loss since birth	Acidosis
	Hyper pigmentation of axilla, aereola, scrotum
	Dehydration and possibly shock

Frequency

The genetic frequency of classical CAH is 1 in 15,000 births. It is an autosomal recessive disorder affecting males and females in equal numbers. It occurs with varying frequency in different ethnic groups.

Treatment

All forms of CAH are managed by replacing the missing hormones with steroids to regulate hormone levels. The treatment will be lifetime.

Next Essential Steps:

1. Contact family for immediate examination of the infant and evaluation of her/his clinical condition.

2. Initiate lab work: IMMEDIATE serum electrolytes and 17-OHP level. The screening program will instruct you in obtaining the 17-OHP testing.

A. If child is ill regardless of electrolyte results:	**B. If child appears well and the electrolytes are abnormal:**	**C. If child appears well:**
Admit to the hospital for further evaluation and fluid management.	Admit to the hospital for further observation and management.	Wait for the electrolyte results. If the electrolytes are normal, observe and wait for the 17-OHP results.

3. Refer to a pediatric endocrinologist as soon as possible for further guidance regarding additional testing that may be indicated and assistance with management of the infant.

Pediatric endocrinologists available for consultation:

Dr. David L. Donaldson Dr. Carol M. Foster Dr. Carol M. Murray Dr. Marie F. Simard	Utah Diabetes Center 615 Arapeen Dr Ste. 100 Salt Lake City, UT 84108 (801) 581-7761
Dr. Mike T. Swinyard	1268 W. South Jordan Parkway Ste. 300 South Jordan, UT 84095 or 5495 S 500 E Ste. 120 Ogden, UT 84405 (801) 838-9090

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Newborn Screening Program